

ABSTRACT OF THE INVENTION

This invention relates to polymorphisms in the IRF6 gene which are associated with various diseases, such as Van der Woude syndrome, Popliteal syndrome, isolated cleft lip and/or palate, and other orofusial clefting disorders. The invention also relates to the 5 nucleotide sequences of this gene, isolated nucleic acids comprising these nucleotide sequences, and isolated polypeptides or peptides encoded thereby. The invention further relates to vectors and host cells comprising the disclosed nucleotide sequences, or fragments thereof, as well as antibodies that bind to the encoded polypeptides or peptides. In addition the invention relates to methods and compositions employing the disclosed 10 nucleic acids, polypeptides or peptides, or antibodies for use in diagnostics and therapeutics for IRF6 associated disorders, e.g., Van der Woude syndrome, Popliteal syndrome, isolated cleft lip and/or palate, and other orofusial clefting disorders.